

Birth Defects: Trisomy 21 Prevalence

Type of EPHT Indicator	Health Outcome
Measures	<p>Prevalence per 10,000 live births over a five year period of Trisomy 21 to mother is two age groups:</p> <ul style="list-style-type: none"> • Less than 35 Years of Age • Greater than or equal to 35 Year of Age
Derivation of measure	<p>Denominator is composed of all live-born infants in geographic region of interest during a calendar year.</p> <p>Numerator is composed of all live-born infants, fetal deaths (where available), and terminations (where available) with Trisomy 21 in the geographic region of interest, during a calendar year.</p> <p>For states that ascertain fetal deaths and/or terminations, two sets of birth prevalence estimates are to be calculated for each birth defect -- one including and one excluding fetal deaths and/or terminations.</p> <p>Diagnosis of cases may be made up to one year of age – ascertainment may be at any time.</p>
Unit	Defect presence at birth (or fetal death/termination)
Geographic Scope	Iowa
Geographic Scale	State, county
Time Period	2001-Most Recent Year Available
Time Scale	5-year period
Rationale	<p>Birth defects pose a significant public health problem. One in 33 babies is born with a structural birth defect in the United States. Birth defects are a leading cause of infant mortality and responsible for considerable morbidity and disability with enormous economic and social costs. A lifetime of medical care and special education for a single child can cost more than \$500,000.</p> <p>Approximately 60% of birth defects are of unknown etiology. The ambient environment remains a source of great public concern, but few environmental exposures have been well-studied. Most birth defects will likely be explained by a complex interaction between genetic predispositions and environmental factors. However, before to the ability to conduct studies to explore these interactions is achieved, linking birth defects-outcome data with environmental hazard or exposure data is critical. The first step in effecting successful linkages of these data is the existence of high quality birth defects prevalence data for which the geospatial and temporal patterns and distributions can be monitored. The environmental public health tracking (EPHT) initiative is well-positioned to bring together birth prevalence data from its state partners to begin analyses of these patterns, which will provide important clues to public health officials and researchers.</p>
Use of the Measure	<p>State</p> <p>Allow for consistent and rapid method for calculating and displaying (using GIS) prevalence at selected geographical areas (i.e, county</p>

	<p>level).</p> <p>Allow for a better understanding of spatial and temporal patterns of selected birth defects.</p> <p>National</p> <p>Allow for comparison of birth prevalence across states which can be used to target interventions. Any comparison of birth prevalence, however, will need to account for the variability in data collection methods between state surveillance systems. (See "Limitations of Data Sources" below and introductory text in appended team recommendations).</p>
Limitations of the Measure	<p>Ideally, incidence rates would be used instead of birth prevalence to measure birth defects occurrence. The numerator of the incidence would be the number of new cases of birth defect A in an area and time period and the denominator would be the number of conceptions at risk of developing birth defect A in that area and time period. Because the both the number of conceptions is unknown and the number of cases "lost" through spontaneous abortions (as well as terminations and later fetal losses depending on the source of ascertainment for the specific surveillance system), incidence cannot be calculated. Birth prevalence is the only appropriate measure that can be reported for birth defects occurrence.</p> <p>It is not feasible, at this time, to recommend that individual-level birth defects surveillance data be made available on even a secure national portal. Most states have strict guidelines with respect to confidentiality and even the publication of birth prevalence data based on <5 cases in a geographic region is generally not done.</p>
Data Sources	<p>State birth defects surveillance systems: The data sources that contribute to birth defects surveillance systems include the following (this varies by system type):</p> <ul style="list-style-type: none"> • Vital records • Hospital records (discharge summaries or disease indices, nursery logs, NICU logs) • Administrative databases (Medicaid, state hospital discharge, HMO) • Specialty data sources (specialty clinics, programs for children with special health care needs) • Prenatal diagnostic centers or genetics clinics • Clinical examination • Local or national laboratories for cytogenetic testing <p>Denominator data will come from state vital records – number of live births, by year, by maternal age, and race/ethnicity. These data may be aggregated and provided to the birth defects surveillance system for calculating birth prevalence, or may be made available on an individual level to the birth defects surveillance system. This varies by state.</p>
Limitations of Data Sources	<p>All states in the US do not have a birth defects surveillance program. Among those that do, there is significant variability between surveillance systems. These include:</p>

- Ascertainment method (active, passive, passive with follow-up/verification)
 - Primary differences are with data sources, coding, availability of verbatim description, and case verification
- Ascertainment of spontaneous fetal deaths and variability in gestational age for inclusion.
- Ascertainment of prenatally diagnosed cases and elective terminations
- Case definitions
- Classification as isolated, multiple or syndromic

Data for specific birth defects may not be collected by each state or may only have been collected recently, limiting historical data for that birth defect.

Address data tends to be address at delivery not conception (more relevant time period for birth defects-related exposure).

Approximately 50% of birth defects surveillance systems do not geocode their address data.